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CASE STUDY

A RARE CASE REPORT OF NOONAN SYNDROME IN A 21 YEAR OLD MALE PATIENT

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ABSTRACT

Noonan syndrome is a rare autosomal dominant disorder with an estimated incidence between one in 1000 to 2500 live births. Characteristic features involve short stature, cardiac defects, haematological problems, developmental delays and several malformations in the oral cavity. Several features of Noonan syndrome resemble those of Turner's syndrome; however a clear demarcation can be made based on the karyotyping of the individual. A child with Noonan syndrome requires additional care and support while carrying out his daily affairs. Early detection of the syndrome can prove to be extremely useful for the child, thereby opting for a multidisciplinary management. The present case report sheds light on the various features associated with Noonan syndrome, thereby helping its early detection. Management of the oral diseases in such patients at the earliest proves beneficial to the child's physical and psychological state. Signs and symptoms generally lessen with age.

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INTRODUCTION

Noonan syndrome was first recognized as a unique entity by Noonan and Ehmke in 1963 as a multisystem disorder, characterized by clinical features such as short stature, hypertelorism, ptosis and low-set ears. (Noonan and Ehmke, 1963) Noonan syndrome is also known as "pseudo-Ullrich Turner syndrome", "Turner like syndrome more details", "female pseudo-Turner syndrome" and "webbed neck syndrome". (Allanson, 1987) Four disease-causing genes (*PTPN11, SOS1, RAF1,* and *KRAS*) have been linked to causing Noonan syndrome along with mutations in the *RASMAPK* signalling pathway. (Bertelloni *et al*., 2013)

Case Report

A 21 year old deaf and mute male patient reported with a chief complaint of pain and swelling in the lower right and left back region of the mouth. He was the third child to healthy parents with a consanguineous marriage. The boy was born full term with extremely low birth weight. His medical history revealed that he had undergone ocular lens surgeries 10 years back.

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The patient's height and weight were 145 centimetres and 29.9 kilograms, respectively. On clinical examination the patient presented with dysmorphic features like microcephaly, small mandible, short stature, hypertelorism, low set posterior ears, mild scoliosis and webbing of the neck. Mental retardation was also present. Restricted spreading of fingers and kolionychia was seen with the feet. Absence of secondary sexual characteristics, hypogonadism, widely spaced nipples and absence of chest/axillary hair was noted. Presence of Café au lait spots (2cm x 1cm) was noted on the trunk. (Fig. 1-8) Oral examination (Fig. 9-11) revealed that the patient had severely compromised oral hygiene. The upper centrals (11, 21) and the lower right 1st permanent molar (46) were absent. Root pieces were seen with all the maxillary teeth except the left lateral incisor (22) and the canine (23). The lower arch seemed to be considerably intact with minor carious lesions in the anterior teeth. The area of chief complaint revealed the presence of infected root pieces with lower left 1st and 2nd permanent molars (36, 37) and lower right 2nd permanent molar (47). Radiographic examination revealed the presence of periapical abscess with the lower permanent molar. Extraction with all the root pieces, RCT with 22 and restoration with 23, 31, 41, 43 and 44 was planned. The patient was scheduled for treatment under general anesthesia owing to his uncooperative behaviour. Before carrying out the procedure, his Pre-Anesthetic Checkup was done along with the required ENT, ophthalmic and endocrine consults. The blood profile reports

showed mild thrombocytopenia and low testosterone levels. All the other reports came in normal and well within the acceptable range. The patient also showed a normal karyotype of 46XY. Based on the scoring scale by Ineke van der Burgt (Van der Burgt *et al*., 1994) and on the karyotyping results, a definitive diagnosis of Noonan's syndrome was established. The required treatment was carried out and the necessary post operative care established. Optimal parent and patient satisfaction was obtained and the patient at present is asymptomatic with relation to any pain or discomfort (Fig.12-14). The patient is now scheduled to undergo hormonal therapy with adjunct grown hormones.

Fig. 1. Short stature and lean weight of the patient

Fig. 2. Low set posterior ears

Fig. 3. Microcephaly and anterior open bite

Fig. 4. Restricted spreading of fingers

Fig. 5. *Kolionychia* seen with toe nails

Fig. 6. Webbing of the neck

Fig. 7. Café au lait spots

Fig. 8. Widely spaced nipples, absence of chest hair

Fig. 9. Pre-operative intro-oral view of teeth in occlusion

Fig. 10. Pre-operative intra-oral view of the maxilla

Fig. 11. Pre-operative intra-oral view of the mandible

Fig. 12. Post-operative intra-oral view of teeth in occlusion

Fig. 13. Post-operative intra-oral view of maxilla

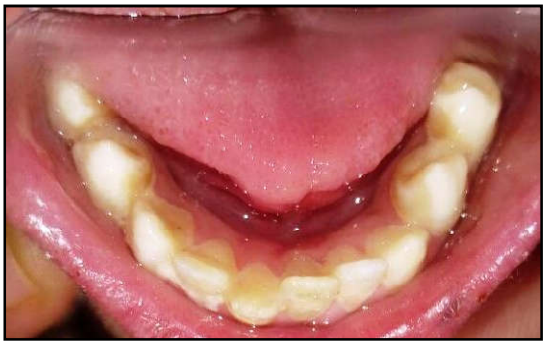

Fig. 14. Post-operative intra-oral view of mandible

DISCUSSION

Noonan's syndrome is a developmental disorder showing autosomal dominant traits. In approximately 50% of the patients with definite NS, a missense mutation is found in the *PTPN11* gene on chromosome 12. (Tartaglia *et al*., 2001) However the failure to identify a PTPN11 mutation doesn't always rule out Noonan syndrome. The diagnosis of Noonan's syndrome can be made on the basis of the Scoring system given by Bergt *et al*. and the Karyotyping. (Van der Burgt *et al*., 1994)

Feature	A = Major	B = Minor
Facial	Typical face dysmorphology	Suggestive face dysmorphobgy
Cardiac	Pulmonary valve stenosis, HOCM and/or ECG typical of NS	Other defect
Height	<P3*	<P10*
Chest wall	Pectus carinatum/excavatum	Broad thorax
Family history	First degree relative with definite NS	First degree relative with suggestive NS
Other	Mental retardation, cryptorchidism and lymphatic dysplasia	One of mental retardation, cryptorchidism, lymphatic dysplasia

Fig. 15. Scoring system for diagnosis of Noonan sundrome

HOCM: hypertrophic obstructive cardiomyopathy;

*P3 and P10 refer to percentile lines for height according to age, with the normal range of variation defined as P3-P97 inclusive

Definitive NS: 1 "A" plus one other major sign or two minor signs; 1 "B" plus two major signs or three minor signs. Owing to the difference in the phenotype and gender predilection, Noonan syndrome can be easily differentiated from Turner's. Then there are a group of distinct syndromes with partially overlapping phenotypes in which causative mutations are found in genes of the RAS-MAPK pathway. These include Cardio-Facio-Cutaneous (CFC) syndrome, Costello syndrome, Neurofibromatosis type 1 (NF1) and LEOPARD syndrome. (Gelb and Tartaglia, 2006) Oral manifestations such as high arch palate, severe dental caries, radicular anomalies of primary molar, malocclusion, micrognathic mandible, early exfoliation of primary canines, labial hypotonia, gingival inflammation, proclined incisors, supernumerary teeth, etc. have profoundly been associated with Noonan syndrome. (Barberia Leache *et al*., 2003; Okada *et al*., 2003) Noonan is also characterized by short stature, mental retardation, cardiac anomalies and dysmorphic facial features. A patient with a suspected case of Noonan must undergo several tests and investigations including; Total blood count, coagulation profile, Karyotyping, complete cardiac examination, audiologic and visual investigations. The treatment for patients with Noonan syndrome is multidisciplinary in nature. No specific pharmacologic therapy is necessary for such patients. Aspirin should be avoided in patients with platelet disorders. Heavy outdoor activities should be restricted in patients with cardiac anomalies. Growth hormone has been used to accelerate growth in some patients with this syndrome. A continuous follow up is required for all the patients to prevent worsening of the prevalent symptoms. Ascertaining the diagnosis of Noonan can be difficult, particularly in adulthood, because of the diverse presentations of the phenotype, which may even become less pronounced with increasing age. (Allanson *et al*., 1985) Antenatal diagnosis proves to be extremely useful. Noonan syndrome should be considered in all foetuses with polyhydramnion, pleural effusions, oedema and increased nuchal fluid with a normal karyotype. (Nisbet *et al*., 1999) Early identification and management of Noonan syndrome is very important. The future of GH therapy in NS remains difficult to predict and additional research is required in this area.

Conclusion

Noonan Syndrome is a rare disorder presenting itself with several deformities. These children usually display a wide array of health problems, making it important for all the practitioners to intervene at the earliest. The pediatric dentist should work diligently to provide the child with comprehensive oral health care along with coordinating with the other team members for effective multidisciplinary management.

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